WASHINGTON STATE Board of Health
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Washington State Department of Health

NEWBORN SCREENING PROGRAM
***SYNOPSIS*: NATIONAL RECOMMENDATIONS**
Report to the Board—October 12, 2005

In response to emerging issues in newborn screening related to advancing medical knowledge and technology, the U.S. Health Resources and Services Administration (HRSA) commissioned the American College of Medical Genetics (ACMG) to conduct an analysis of the scientific literature on the effectiveness of newborn screening and gather expert opinion to delineate the best evidence for screening specified conditions and develop recommendations focused on newborn screening, including the development of a uniform condition panel. It was expected that the analytical endeavor and subsequent recommendations be based on the best scientific evidence and analysis of that evidence. Among other issues the ACMG was specifically asked to develop recommendations to address a uniform condition panel.

The ACMG report was released in March 2005 in response to the request. The report, Newborn Screening: Toward a Uniform Screening Panel and System is available at http://mchb.hrsa.gov/screening. HRSA solicited public comments on the report through May 2005 but has not yet released information on the comments received.

In the report, 29 conditions are identified as primary targets or core panel conditions for screening. Washington currently screens for 13 of these and two are under current consideration for screening in our state (see listing on page 3). This leaves 16 conditions not screened for or under current consideration.

The report has met some criticism; in particular that much of the weight of evidence was dependent on expert opinion as opposed to observational or clinical trials. Nonetheless, as the first national recommendation for a uniform panel, the report marks a major milestone in newborn screening and its recommendations are supported by many organizations including the National March of Dimes, the American Academy of Pediatrics, and Save Babies through Screening, a national organization of parent advocates.

In December 2004 the Board of Health requested that after publication of the report, the Department of Health review it to determine: if there are effective interventions for the 16 new conditions; a preliminary assessment of associated costs; Washington's capacity to detect and treat the disorders; and the number of newborns in Washington who could potentially be identified with the conditions.

The Department has reviewed the report as requested and finds that while there are interventions for each of the disorders there is a large range in the effectiveness of treatment between the disorders.

The cost of screening for them would be minimal (probably less than $1 per child for all 16) because they can easily be included within the same analytical process (tandem mass spectrometry) used for other disorders in the screening battery. The specialty clinic that treats

children with these disorders has indicated that expanded infrastructure is being developed to care for children with the five disorders added in 2004. With this expansion there will be sufficient capacity, at this time, to care for infants diagnosed with these new disorders without additional funding. However, it must be noted that the long-term funding mechanism for the clinic is being reviewed. A temporary fee that is charged through birthing hospitals is funding the current infrastructure expansion, but whether this is an appropriate mechanism for continued funding is an ongoing policy issue.

Because the conditions are uncommon, and many have only been known for a few years, their frequency is not well established. The report lists them as either greater than 1 in 75,000 births or less than one in 100,000 births. Using these numbers as point estimates (i.e. assuming that the frequencies are either exactly 1 in 75,000 or 1 in 100,000) we would expect to identify perhaps 14 affected infants among 76,000 infants screened each year if we screened for all 16 additional conditions. Given the uncertainty, the actual numbers are almost certain to vary.

In summary, despite some shortcomings, the substantial body of work reflected in the report presents a challenge for consideration by newborn screening programs across the country. The Department of Health's review of the recommendations finds that at least some of the 16 new conditions may meet the Board's criteria and be good candidates for addition to our state's newborn screening panel.

Disorders included in the American College of Medical Genetics recommendations for a uniform newborn screening panel panel:

Currently included in Washington's panel

1. Biotinidase deficiency
2. Congenital adrenal hyperplasia
3. Congenital hypothyroidism
4. Galactosemia (GALT)

   Hemoglobinopathies:
   5. Sickle cell anemia
   6. Hb S/Beta-thalassemia
   7. Hb S/C disease

8. Homocystinuria
9. Maple syrup urine disease (MSUD)
10. Medium chain acyl-CoA dehydrogenase (MCAD) deficiency
11. Phenylketonuria (PKU)

Under current consideration by the Board

12. Hearing loss
13. Cystic fibrosis (CF)

Remaining (all are detected through MS/MS screening of dried blood spot)

14. 3-Methylcrotonyl-CoA carboxylase deficiency
15. 3-OH 3-CH3 glutaric aciduria
16. Argininosuccinic acidemia
17. Beta-ketothiolase deficiency
18. Carnitine uptake defect
19. Citrullinemia
20. Glutaric acidemia type I
21. Isovaleric academia
22. Long-chain L-3-OH acyl-CoA dehydrogenase deficiency
23. Methylmalonic acidemia
24. Methylmalonic acidemia (mutase deficiency)
25. Multiple carboxylase deficiency
26. Propionic acidemia
27. Trifunctional protein deficiency
28. Tyrosinemia type I (TYR I)
29. Very long-chain acyl-CoA dehydrogenase deficiency